

CLAIMS:

1. A DNA analysis system which includes a unit that effects both extraction of DNA and amplification by identical replication of a region of interest of extracted DNA strands, with a proteinase, as defined, being used in the unit at least to effect extraction
5 of DNA.
2. The system of claim 1 in which the amplification includes nucleotide sequence detection for the purpose of looking for specific sequences of DNA.
- 10 3 The system of claim 2 in which the unit includes an attached fluorimeter and light source.
4. A DNA analysis system which includes:
 - a thermal cycler operable as an extraction stage for extracting DNA from a sample to be tested and as an amplification stage for replicating identically a region of interest in DNA strands extracted from the sample, a proteinase, as defined, being used
15 in the thermal cycler at least in the extraction stage;
 - a purification stage for purifying the amplified material from the thermal cycler;
 - and
- 20 an analysis stage for analysing the purified sample to obtain genetic information relating to the sample.
5. The system of claim 4 in which the analysis stage comprises a separation stage and a detection stage.
- 25 6. The system of claim 4 or claim 5 which includes a sequencing stage preceding the analysis stage.
- 30 7. The system of claim 6 in which the thermal cycler is used for the sequencing stage.
8. The system of claim 6 or claim 7 in which the purification stage incorporates a size filtration matrix comprising a gel filtration media incorporating a filtering resin, the matrix allowing larger fragments of DNA through from the amplification stage
35 before any smaller fragments and other unwanted substances.

9. The system of claim 8 in which the larger fragments are collected for use in the sequencing stage.
10. The system of claim 9 in which the sequencing stage tags ends of the fragments with dideoxynucleoside triphosphates (ddNTP's) labelled with different fluorochromes before grading.
11. The system of claim 10 in which the grading forms the first step of the separation stage and incorporates separating the fragments into fragments of differing lengths by a separation device.
12. The system of claim 11 in which the separation device is an electrophoresis device.
13. The system of claim 12 in which the electrophoresis device is a capillary electrophoresis device and includes a detector for detecting information relating to tagged fluorescent nucleotides at the end of each of the DNA fragments.
14. The system of claim 13 in which the detector includes a laser device that irradiates the ends of the DNA fragments to cause the fluorescent ends to fluoresce.
15. The system of claim 14 which includes a reader for reading the fluorescent ends of the fragments.
16. The system of any one of claims 4 to 15 in which the thermal cycler includes a controller which controls the various stages of preparation of the sample.
17. The system of claim 16 in which the thermal cycler includes a heating mechanism for heating the sample, contained in one or more vials or test tubes, received in the thermal cycler.
18. The system of claim 17 in which the heating mechanism is controlled by the microcontroller to maintain the sample at the required temperatures at the various stages of extraction, amplification and sequencing.

19. The system of claim 17 or claim 18 which includes a dispensing device for depositing the material to be analysed in the thermal cycler.

20. The system of claim 19 in which the thermal cycler includes a holder for
5 holding replacement tips for the dispensing device.

21. The system of claim 20 in which the holder is arranged on the thermal cycler adjacent the heating mechanism within reach of the range of movements of the dispensing device.

10 22. The system of claim 21 in which the holder includes reservoirs for various solutions adjacent the replacement tips.

15 23. The system of any one of claims 20 to 22 in which the purification stage is mounted on the holder adjacent the heating mechanism of the thermal cycler.

24. The system of any one of claims 4 to 23 which includes a monitoring means for monitoring the analysis stage.

20 25. The system of claim 24 in which the monitoring means is in the form of a computer having a display on which data relating to the analysed sample are displayed.

26. A method of preparing a sample for DNA analysis, the method including the step of using a single unit to effect both extraction of DNA and amplification by
25 identical replication of a region of interest of extracted DNA strands, with a proteinase, as defined, being used in the unit at least to effect extraction of DNA.

27. The method of claim 26 which includes the step of looking for specific sequences during amplification by including nucleotide sequence detection in the
30 amplification stage.

28. The method of claim 27 which includes performing nucleotide sequence detection during amplification by adding fluorescently labelled oligonucleotides that can target a specific sequence of DNA.

29. The method of claim 28 which includes using a thermal cycler that has an attached fluorimeter and light source.

30. A method of preparing a sample for DNA analysis, the method including the
5 steps of:

placing a sample of material to be analysed in a thermal cycler and adding a predetermined quantity of proteinase to the thermal cycler;

cycling the mixture through a predetermined temperature profile to effect extraction of DNA material from the sample;

10 in the thermal cycler, subjecting the extracted DNA material to an amplification stage replicating identically a region of interest in the extracted DNA material; and sequencing the amplified material.

31. The method of claim 30 which includes sequencing the material by a dideoxy
15 method of sequencing which includes the steps of sequencing, separation and detection.

32. The method of claim 30 or claim 31 which includes, as part of separating the DNA material, purifying the material and sequencing the purified DNA material.

20 33. The method of claim 32 which includes effecting the sequencing of the purified DNA material for separation and detection using the thermal cycler.

34. The method of claim 32 or claim 33 which includes purifying the material by passing the material through a size filtration matrix comprising a gel filtration media
25 incorporating a filtering resin, the matrix allowing larger fragments of DNA through from the amplification stage before any smaller fragments and other unwanted substances.

35. The method of claim 34 which includes collecting the larger fragments for use
30 in the sequencing of the material.

36. The method of claim 35 which includes tagging ends of the fragments with dideoxynucleoside triphosphates (ddNTP's) labelled with different fluorochromes before grading.

37. The method of claim 36 in which the grading forms the first step of the separation stage and the method incorporates separating the fragments into fragments of differing lengths.

5 38. The method of claim 36 or claim 37 which includes detecting information relating to tagged fluorescent nucleotides at the end of each of the DNA fragments.

39. The method of claim 38 which includes irradiating the ends of the DNA fragments to cause the fluorescent ends to fluoresce and reading the fluorescent ends of
10 the fragments.

40. A purification stage for a DNA analysis system, the purification stage including a conduit; and
a gel filtration medium contained in the conduit, the gel filtration medium being
15 a resin of microscopic, synthetic beads.

41. The purification stage of claim 40 in which the gel filtration medium is of microscopic beads synthetically derived from a polysaccharide.

20 42. The purification stage of claim 41 which includes a control device for controlling the passage of the sample through the conduit.

43. A method of purifying a DNA sample, the method including the step of passing the sample through a conduit containing a gel filtration medium in the form of a resin
25 of microscopic, synthetic beads to effect purification of the sample.

44. The method of claim 43 which includes forming the beads from a polysaccharide.

30 45. The method of claim 43 or claim 44 which includes controlling the passage of the sample through the conduit.

46. A DNA analysis system which includes:
a unit operable at least as an extraction stage for extracting DNA from a sample
35 to be tested and as an amplification stage for replicating identically a region of interest in DNA strands extracted from the sample;

a microfluidic device mounted on the unit and defining a plurality of wells interconnected by a channel, a sample undergoing various stages of preparation being moved sequentially from one well to another via the relevant interconnecting channel; and

5 a control arrangement for controlling movement of the sample between said wells.

47. The system of claim 46 in which the unit also operates as a sequencing stage.

10 48. The system of claim 46 or claim 47 in which the control arrangement includes an electric field generating means that moves a charged solution between the wells through the channels.

15 49. The system of claim 48 in which the electric field generating means comprises a plurality of electrodes, each of said predetermined wells having an electrode associated with it.

50. The system of any one of claims 46 to 49 in which at least certain of the wells operate as waste wells in which waste material, separated out from the sample, is deposited for disposal.

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51. The system of any one of claims 46 to 50 which includes a dispensing arrangement for depositing reagents in the wells.

25 52. The system of claim 51 in which the dispensing arrangement comprises at least one pipette for dispensing the reagents.

53. A method of preparing a sample for DNA analysis, the method including the steps of:

30 placing a sample of material to be analysed in a first well of a microfluidic device having a plurality of wells interconnected by channels;

 effecting a first preparatory stage in the first well of the device;

 controlling movement of the sample from one well, sequentially, to further wells in the microfluidic device and carrying out further preparatory stages at each of

35 predetermined wells in the device.

54. The method of claim 53 which includes modifying an existing thermal cycler by mounting the microfluidic device on the thermal cycler.
55. The method of claim 53 or claim 54 which includes controlling the movement of 5 the sample from well to well by means of an electric field generating means that moves a charged solution between the wells through the channels.
56. The method of claim 55 which includes associating an electrode with each well and controlling the movement of the sample between wells by changing the potential of 10 the wells relative to one another.
57. The method of any one of claims 53 to 56 which includes designating one of the wells as a waste well and depositing waste material, separated out from the sample, in the waste well.